

Patent claims

1. Nucleic acid, which includes

- a) the nucleotide sequence shown in Fig.1 or a protein-coding segment thereof,
- b) one of the sequence from a) within the context of the degeneration of the genetic code corresponding nucleotide sequence,
- c) one with the sequences from a) and/or b) under stringent conditions

hybridizing nucleotide sequence, except for the EST sequences:

AA165403, AA455594, AA314472, N34087, AA452340, AA182700,
N41615, AA470049, AI751597, AA463289, AA132459, W31487,
R56355, H58271, H16122, W77956, AA193332, AA323923, AA370209,
AA296758, W72757, AA093971, AA385544, AA386175, AA165402,
AW085713, H42806, AA093977, AI161152, AA370011, AI671702,
R71215, AA885343, T79297, AI814869, R81567, AI082713, N29615,
AW087726, AW075710, AI952608, AI818073, AI784445, AI432812,
AI375568, AI372904, AI364106, AI143379, AA993074, AA953985,
AA862385, AA761084, AA576229, AA569223, AA463198, AA452117,
AA416877, AA074872, W16851, W04568, N40176, AW068354,
AA857004, H58663, H15819, AW264944, AI923965, AI692214,
AI475321, AI435987, AA961068, AA206059, AI469161, T84789,
AA507257, AA707515, AA132458, AA179262, T79211, W31505,
N25699, T99574, T99363, AI751598, AA713668, T91119, AW105515,
AA370208, AI422128, R81568, AI038899, AI971847, AI540650,
AI826106, AA885960, R56263, AA825431, T99147, D31503 and
AF049564, or

- d) a complementary sequence to the sequences of a) and/or b).

2. Nucleic acid according to claim 1, which includes a protein-coding segment comprising of preferably at least 30 nucleotides of the nucleotide sequence shown in Fig. 1.

3. Nucleic acid, which shows a homology of more than 65% with the nucleotide sequence according to claim 1 or a segment thereof.

Sub
a)

- Sub A2
4. Modified nucleic acid or nucleic acid analogue, which includes a nucleotide sequence according to one of the claims 1 to 3.
5. Recombinant vector, which includes at least one copy of a nucleic acid according to one of the claims 1 to 3 or a section thereof.
6. Recombinant vector according to claim 5, which enables the expression of the nucleic acid in a suitable host cell.
- Sub A3
- 10 7. With a nucleic acid according to one of the claims 1 to 3 or a vector according to claim 5 or 6 transformed cell, a corresponding non-human transgenic organism or animal models, which stably produce (knock-in) the product of the nucleic acid according to one of the claims 1 to 3 or whose corresponding natural gene was destroyed deliberately (knock-out).
- 15 8. Polypeptide or a salt thereof, which is coded by a nucleic acid according to one of the claims 1 to 3.
9. Polypeptide according to claim 8, which exhibits
- a) the amino acid sequence shown in Fig. 2 or
- 20 b) a homology of more than 60% with the amino acid sequence shown in Fig. 2 or a salt thereof.
- Sub A4
- 25 10. Fragment of the polypeptide according to claims 8 or 9 with at least 100 amino acids or salts thereof.
11. Modified polypeptide, which includes an amino acid sequence according to claims 8 or 9.
12. Methods for the synthesis of the polypeptide according to claim 8 or 9, which
- 30 includes the cultivation of cells according to claim 7 as well as the isolation of the polypeptide according to claim 8 or 9.
13. Use of a polypeptide according to claim 8 or 9 or of fragments of this polypeptide as an immunogen for the production of antibodies.

14. Antibodies against a polypeptide according to claim 8 or 9.
15. Method for the identification of effectors of a protein according to claim 8 or 9,
5 with the help of which various potential effector substances can be tested on cells,
which express the protein.
16. Pharmaceutical composition, which includes as active component
a) a nucleic acid according to one of the claims 1 to 4,
10 b) a vector according to claim 5 or 6,
c) a cell according to claim 7,
d) a polypeptide according to claim 8, 9, 10 or 11,
e) an antibody according to claim 14
and which contains the pharmaceutically usual carrier, auxiliary and/or additive
15 substances.
17. Use of a composition according to claim 16 for diagnosis of diseases, which are
associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis
and/or tumor progression, or a predisposition to such diseases.
- 20 18. Use of a pharmaceutical composition for diagnosis of diseases which are associated
with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or
tumor progression, or a predisposition to such diseases, which contains as an active
component
25 a) an EST sequence according to claim 1c,
b) a recombinant vector which includes at least one copy of the EST sequences
mentioned above,
c) a recombinant vector according to b) which enables the expression of the
nucleic acid in a suitable host cell,
30 d) a cell according to claim 7, whereas the nucleic acid consists of one of the EST
sequences mentioned above,
e) a polypeptide being coded by one of the EST sequences mentioned above or a
salt thereof or,

- f) a polypeptide according to e) which exhibits the amino acid sequence shown in Fig.2 or a homology of more than 60% with the amino acid sequence shown in Fig.2 or a salt thereof,
- g) a fragment of the polypeptide according to e) or f) with at least 100 amino acids or a salt thereof,
- h) a modified polypeptide which includes an amino acid sequence according to e) or f),
- i) an antibody against a polypeptide according to e) or f) and which contains pharmaceutically usual carrier, auxiliary and/or additive substances.
19. Use of a composition according to claim 16 for the therapy or prevention of diseases, which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumor genesis and/or tumor progression.
20. Use of a pharmaceutical composition according to claim 18 for the therapy or prevention of diseases, which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression.
21. Use of a composition according to claim 16 for a gene therapy of diseases, which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression.
22. Use of a pharmaceutical composition according to claim 18 for gene therapy of diseases which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression.
23. Methods for diagnosing diseases, which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression or a predisposition to such diseases, during which a patient or a sample from the patient is brought in contact with a composition according to claim 16 and the nucleotide sequence and/or the expression of a nucleic acid according to claim 1 is determined.
24. Methods for the therapy or prevention of diseases, which are associated with DNA

repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression, during which a patient is administered a composition according to claim 16, which contains the active components in an amount effective against the disease.

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